Late Prenatal Diagnosis of Hydrometrocolpos Secondary to a Cloacal Anomaly by Abdominal Ultrasonography with Complementary Magnetic Resonance Imaging

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SUMMARY

Objective: Prenatal diagnosis of a cloacal anomaly is difficult. Magnetic resonance imaging (MRI) can assist in the identification of the connection and continuity of a cystic mass to confirm the diagnosis of a cloacal anomaly.

Case Reports: In the first case, a fetal abdominal cystic mass was observed at 32 weeks of gestation. Ultrasonography revealed a retrovesical septate hypoechoic mass with bilateral hydronephrosis. MRI demonstrated a midline cystic mass connected to a dilated uterus and a possible fistula between the bladder and vagina. In the second case, a fetal abdominal septate cystic mass was identified using ultrasonography at 34 weeks of gestation. MRI was performed and demonstrated hydrocolpos/hydrometrocolpos originating from a uterine didelphis with left dysgenesis and a possible vesicovaginal fistula. After birth, both newborns underwent immediate surgical intervention with good outcomes.

Conclusion: MRI facilitated the prenatal diagnosis of cloacal anomalies and allowed additional time for parental counseling and planning of the delivery method with subsequent neonatal intensive care and surgical and urologic consultations. [*Taiwan J Obstet Gynecol* 2008;47(1):79–83]

Key Words: cloacal anomaly, hydrometrocolpos, magnetic resonance imaging

Introduction

Prenatal diagnosis of a cloacal anomaly using ultrasonography (US) remains a clinical challenge. Magnetic resonance imaging (MRI) has been recognized as a useful tool that can be used to identify the connection and continuity of a cystic mass and help make the diagnosis of a cloacal anomaly when US alone is inconclusive [1]. We report herein two cases of cloacal anomalies complicated by hydrometrocolpos and/or bilateral hydronephrosis diagnosed prenatally using US with MRI confirmation. In addition, we reviewed reports in the literature regarding the outcomes of fetal cloacal anomalies.

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Accepted: April 11, 2007

Case Reports

Case 1

A 38-year-old Taiwanese, gravida 3, para 2, was referred to our hospital for evaluation of a fetal abdominal cystic mass observed at 34 weeks of gestation. She had chronic hypertension for years, but the prenatal course had been uncomplicated other than a dilated fetal abdomen first noted at 32 weeks of gestation in the primary obstetrics clinic. US demonstrated a singleton fetus of normal size in the vertex presentation. Within the fetal abdomen, a retrovesical, septate, hypoechoic mass measuring 101 × 63 mm and marked bilateral hydronephrosis were noted (Figure 1). Neither polyhydramnios (amniotic index, 177 mm) nor profuse fetal

ascites was described. Fetal karyotyping was recommended but refused by the mother. MRI was performed to obtain more detailed information of the fetal abdominal mass. Images of the MRI confirmed that the abdominal mass was located in the midline, posterior to the bladder, and showed a connection with the dilated uterine cavity. In addition to bilateral hydronephrosis, a possible fistula formation between the bladder and vagina was also noted (Figure 2). At 36 weeks of gestation, a female infant weighing 3,120 g with Apgar scores of 3 and 6 at 1 and 5 minutes, respectively, was delivered via the abdomen. Abdominal exploration on the third postnatal day revealed a hydrocolpos with vaginal atresia, compatible with a cloacal anomaly and a urogenital sinus. After staging surgeries, a vaginotomy, pigtail drainage of the pelvic cyst, and a flap vaginoplasty were performed via the abdominal-perineal approach.

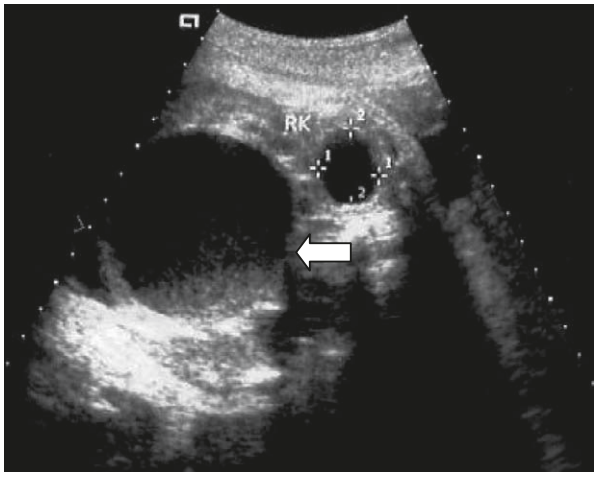

Figure 1. Ultrasonography on transverse view of the lower abdomen of the fetus, with the right kidney (RK) exhibiting hydronephrosis and a cystic abdominal mass (arrow) measuring 101 × 63 mm.

The infant was well with normal development and renal function at the 9-month follow-up examination.

Case 2

A 31-year-old Taiwanese, gravida 1, was referred to our hospital for a fetal abdominal cystic mass observed using US at 31 weeks of gestation. US demonstrated a singleton fetus of normal size in the breech presentation. A septate pelvic cystic mass without hydronephrosis or fetal ascites was noted; the amniotic fluid index was normal. MRI was performed at 34 weeks of gestation, revealing a uterine didelphis with a double vagina, a longitudinal vaginal septum, right hydrometrocolpos, left hydrocolpos (Figures 3 and 4), and a possible vesicovaginal fistula (Figure 5). In addition, a focal dilated sigmoid colon with poor visualization of the rectum, which was consistent with rectal atresia, was also noted (Figure 4). At 36 weeks of gestation, a female infant weighing 2,600 g and with Apgar scores of 8 and 9 at 1 and 5 minutes, respectively, was delivered via the abdomen. Neonatal examination revealed ambiguous genitalia with anal and vaginal atresia. A postnatal abdominal US demonstrated an enlarged double uterus, vagina, bilateral hydronephrosis, and communication between the uterus and urinary bladder. Sigmoid loop colostomy was performed on the first postnatal day, and the infant had regular follow-up examinations without severe complications at the time of this writing, and the subsequent corrective surgery was anticipated to be performed at 3 years of age.

Discussion

Cloacal anomalies derive from the developmental failure of the urorectal fold, which separates the rectum

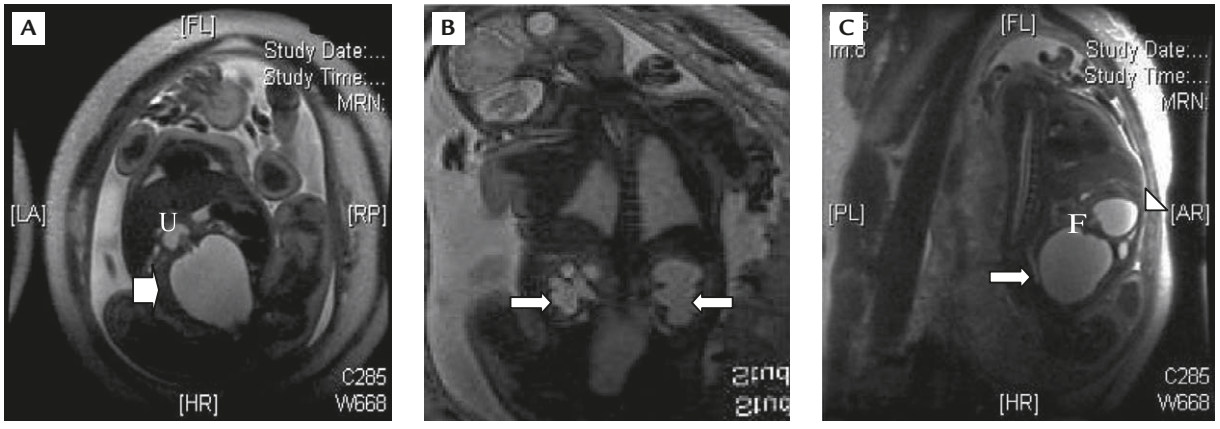

Figure 2. Fetal magnetic resonance imaging at 36 weeks of gestation. T2-weighted image in the coronal view showing: (A) fluid-filled mass (thick arrow) in the midline and connected to the dilated uterus (U); (B) marked bilateral hydronephrosis (arrows); and (C) T2-weighted image in the sagittal view showing the cystic mass (arrow) posterior to the bladder and a possible fistula (F) communicating with the bladder (arrowhead).

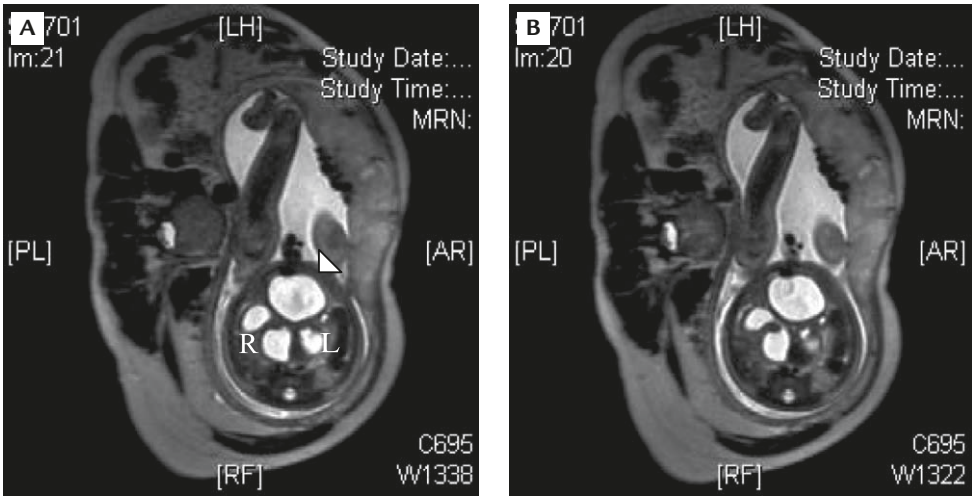

Figure 3. Fetal magnetic resonance imaging at 34 weeks of gestation. T2-weighted image in the transverse view showing: (A) a dilated fetal bladder (arrowhead) in the midline with pelvic cysts posterior to it, indicating right hydrometrocolpos (R) and left hydrocolpos (L); and (B) the communication between dilated vagina and uterine cavity over the right side of the fetus.

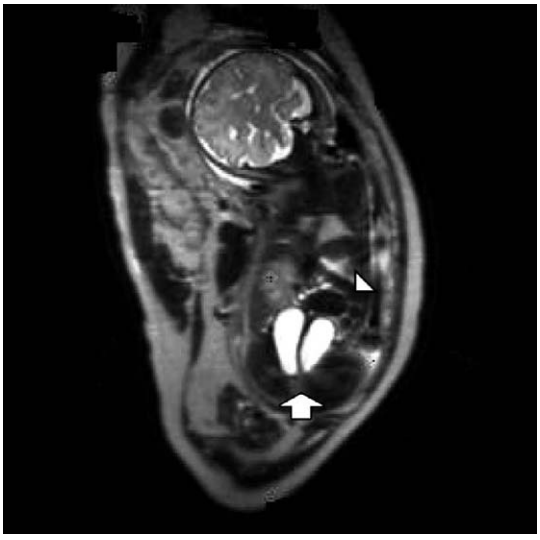

Figure 4. Fetal magnetic resonance imaging at 34 weeks of gestation. T2-weighted image in the coronal view showing focal dilated sigmoid colon (arrowhead) with poor visualization of the rectum and a double vagina with hydrocolpos and a longitudinal septum (thick arrow).

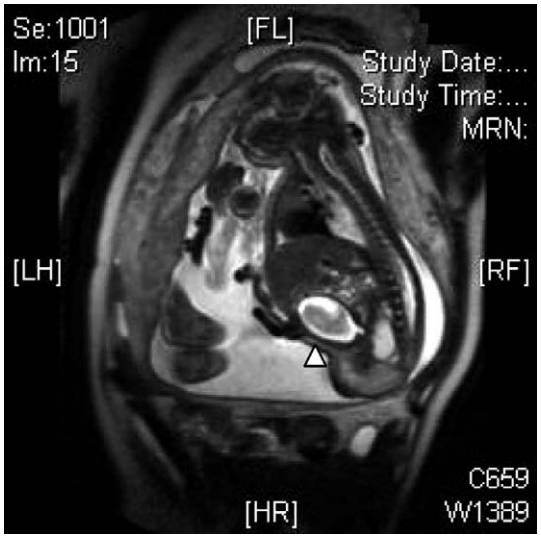

Figure 5. Fetal magnetic imaging at 34 weeks of gestation. A possible fistula from the dilated urinary bladder (arrowhead).

from the uterovaginal tract. It often presents as a common single perineal opening for the genital, urinary and gastrointestinal tracts [2,3]. As urine accumulates in the vagina, a large hydrocolpos develops; and furthermore, as obstruction of the lower urinary tract develops, hydronephrosis may occur.

The incidence of newborn cloacal anomalies is about 1 in 50,000 to 400,000 deliveries [4,5]. The cloaca is a transient structure during embryonic development, which forms from the developing tail fold at 3 weeks of gestational age by the confluence of the allantois and hindgut. By the sixth week of embryonic life, the cloaca is divided, resulting in a urogenital sinus anteriorly and a separate hindgut posteriorly [5]. The urethra subsequently develops from the caudal end of the urogenital sinus after its separation from the cloaca [6].

Development may be arrested at any stage, and a cloacal anomaly can occur as a wide spectrum of diseases involving the genitourinary, rectal, perineal and external genital anatomy [7,8]. Hydrometrocolpos develops in the female fetus as a result of vaginal outflow obstruction, leading to the accumulation of secretions. It might be secondary to the presence of a transverse vaginal septum, vaginal atresia or imperforate hymen [9,10].

The prenatal diagnosis of hydro(metro)colpos has been reported at as early as 25 weeks of gestation [10], and the mean gestational age at the time of diagnosis of cloacal anomalies is reported to be 27 weeks

(range, 19–33 weeks) using US; many cases are diagnosed during the third trimester or after birth [11,12]. Our two cases were both detected using US initially at 32 and 34 weeks' gestation. In the presence of a fetal pelvic cystic mass, however, other abnormalities, including bowel atresia(s), megacystis microcolon, bowel duplication, ovarian cysts and obstructive uropathy, must also be differentiated.

The sequence of events seen on serial prenatal US include transient fetal ascites, appearance and progressive enlargement of a bilobed cystic structure arising from the fetal pelvis, which may contain debris, a poorly visualized fetal bladder, bilateral hydronephrosis, reduction in amniotic fluid volume, growth retardation, and vertebral anomalies [2,3]. The combination and development of these findings, together with a female karyotype, form the basis for the prenatal diagnosis of a cloacal anomaly. Fetal ascites is sometimes detected in the mid-trimester but resolves. Urine escapes from the hydrocolpos via the fallopian tubes into the abdominal cavity [2,3]. Later, tubal occlusion probably occurs because of chronic irritation by urine and meconium. This may result in an increase in the size of the hydrocolpos, further lower urinary tract obstruction, and worsening hydronephrosis [2,3]. Oligohydramnios may also develop as a consequence of urinary tract obstruction, which may lead to pulmonary hypoplasia if it occurs before 24 weeks of gestation [5].

Both of the cases presented herein demonstrated marked pelvic cystic masses, with or without septa and bilateral hydronephrosis. Normal amniotic fluid index, bladder, and fetal size without the entire abovementioned constellation of traits were noted. The cloacal anomalies in our cases may have been of lower severity and continued to drain the fetal bladder well via the cloaca initially. If oligohydramnios occurs, it may be a poor sign for neonatal outcome and termination of pregnancy should be considered.

MRI enables high-quality fetal images to be acquired regardless of the mother's physical condition or fetal position, whereas US may be inconclusive because of maternal obesity, oligohydramnios or suboptimal fetal position [1]. In Case 1, the US demonstrated a fetal pelvic cystic mass with marked bilateral hydronephrosis. With the help of MRI, it not only defined the connection and continuity of the pelvic cyst with the bladder and rectum, but also showed the possible fistulous communication with the bladder, which helped us make a more precise diagnosis prenatally. In Case 2, the MRI also played an important role in our prenatal diagnosis of uterine didelphis with a double vagina and right hydrometrocolpos, in spite of the suggestion of an ovarian tumor initially. The communication between the bladder and uterus or vagina was also differentiated. There was no four-chamber view of the fetal hydrometrocolpos secondary to a cloacal anomaly, similar to that described in a previous report by Hayashi et al [1]. This discrepancy may have been because of unilateral obstruction of the fallopian tube. The subsequent postnatal image study also confirmed the diagnosis of left uterine dysgenesis.

The diagnosis of persistent urogenital sinus may be associated with other congenital malformations, including the genitourinary tract (33%), gastrointestinal tract (13%), and cardiovascular system (13%) [3]. The malformations sometimes overlap with VACTERL association or caudal regression syndrome [13]. It also may present with associated malformations and syndromes, such as McKusick-Kaufman, Ellis-van Creveld or Bardet-Biedl syndromes [14]. It is, therefore, essential to examine the genitalia, spine, and heart in all cases carefully.

The presence of urinary tract obstruction in fetal life may cause long-term adverse sequelae. Fifty percent of patients with clocal abnormalities developed chronic renal failure in childhood, and a mortality rate of 7% from renal failure has been noted [5]. The renal impairment and continence issues [15] must be considered when counseling families prenatally. A higher incidence of urine incontinence (72% *vs.* 28%) was also noted in the group with long common channel (> 3 cm) compared with the group with short common channel (< 3 cm) [16]. A poorer prognosis associated with both lung hypoplasia and significant impaired renal function was also noted when an early second-trimester diagnosis was made [4]. Insertion of a vesicoamniotic shunt may be of benefit in those fetuses with severe oligohydramnios and severe hydronephrosis. The option of terminating the pregnancy should also be discussed with parents when this abnormality is suggested before viability [5].

Until the mid-1950s, cloacal anomalies could not be corrected surgically and caused death [17]. In cases with hydrocolpos, Pena et al [16] first reported the need to not only perform a colostomy, but also drain the dilated vagina to prevent complications. After corrective surgery, the outcomes with respect to urine incontinence, sexual function, and pregnancy were quite good [14]. In our two cases, one of the neonates underwent staging surgeries for drainage and vaginoplasty, and the other neonate underwent a colostomy by a pediatric surgeon. Both neonates have had uncomplicated developmental courses thus far.

Counseling of the parents whose fetus is diagnosed prenatally as having a cloacal abnormality should include a discussion regarding possible anomalies, as well as

possible complications of labor. In a case with such anomalies, delivery should be performed at a center where integrated perinatal, neonatal and surgical services are available.

In conclusion, when a fetal pelvic cystic mass is detected using US with female genital anatomy or karyotyping, fetal anomalies including a cloacal anomaly should be considered, and one should look for cardiac and vertebral anomalies more carefully. MRI can be a useful tool, along with abdominal US, to assess such fetal anomalies.

References

1. Hayashi S, Sago H, Kashima K, et al. Prenatal diagnosis of fetal hydrometrocolpos secondary to a cloacal anomaly by magnetic resonance imaging. *Ultrasound Obstet Gynecol* 2005;26:577–9.
2. Cilento BG Jr, Benacerraf BR, Mandell J. Prenatal diagnosis of cloacal malformation. *Urology* 1994;43:386–8.
3. Zaccara A, Gatti C, Silveri M, et al. Persistent cloaca: are we ready for a correct prenatal diagnosis? *Urology* 1999; 54:367.
4. Ohno Y, Koyama N, Tsuda M, Arii Y. Antenatal ultrasonographic appearance of a cloacal anomaly. *Obstet Gynecol* 2000;95:1013–5.
5. Warne S, Chitty LS, Wilcox DT. Prenatal diagnosis of cloacal anomalies. *BJU Int* 2002;89:78–81.
6. Liang X, Ioffe OB, Sun CC. Cloacal dysgenesis sequence: observations in four patients including three fetuses of second trimester gestation. *Pediatr Dev Pathol* 1998;1:281–8.
7. Lande IM, Hamilton EF. The antenatal sonographic visualization of cloacal dysgenesis. *J Ultrasound Med* 1986;5:275–8.
8. Gaucherand P, Vavasseur-Monot C, Ollagnon E, Boisson C, Labaune JM, Basset T, Yared G. McKusik-Kaufman syndrome: prenatal diagnosis, genetics and follow up. *Prenat Diagn* 2002;22:1048–50.
9. Chen CP, Liu FF, Jan SW, Chang PY, Lin YN, Lan CC. Ultrasound-guided fluid aspiration and prenatal diagnosis of duplicated hydrometrocolpos with uterus didelphys and septate vagina. *Prenat Diagn* 1996;16:572–6.
10. Winderl LM, Silverman RK. Prenatal diagnosis of congenital imperforate hymen. *Obstet Gynecol* 1995;85:857–60.
11. Mirk P, Pintus C, Speca S. Ultrasound diagnosis of hydrocolpos: prenatal findings and postnatal follow-up. *J Clin Ultrasound* 1994;22:55–8.
12. Odibo AO, Turner GW, Borgida AF, Rodis JF, Campbell WA. Late prenatal ultrasound features of hydrometrocolpos secondary to cloacal anomaly: case reports and review of the literature. *Ultrasound Obstet Gynecol* 1997;9:419–21.
13. Winter RM, Baraitser M. *London Dysmorphology Database*. Oxford: Oxford University Press, 2000.
14. Geipel A, Berg C, Germer U, Ahrens P, Gloeckner-Hofmann K, Moller J, Gembruch U. Diagnostic and therapeutic problems in a case of prenatally detected fetal hydrocolpos. *Ultrasound Obstet Gynecol* 2001;18:169–72.
15. Brock WA, Pena A. Cloacal abnormalities and imperforate anus. In: Kelalis PP, King LR, Belman AB, eds. *Clinical Pediatric Urology*, 3rd edition. Philadelphia: WB Saunders, 1992:920–42.
16. Pena A, Levitt MA, Hong A, Midulla P. Surgical management of cloacal malformations: a review of 339 patients. *J Pediatr Surg* 2004;39:470–9; discussion 470–9.
17. Jones PG. Exomphalos: a review of 45 cases. *Arch Dis Child* 1963;38:180–7.